



CTNS gene

cystinosin, lysosomal cystine transporter

Normal Function

The *CTNS* gene provides instructions for making a protein called cystinosin. This protein is located in the membrane of lysosomes, which are compartments in the cell that digest and recycle materials. Proteins digested inside lysosomes are broken down into smaller building blocks, called amino acids. The amino acids are then moved out of lysosomes by transport proteins. Cystinosin is a transport protein that specifically moves the amino acid cystine out of the lysosome.

Health Conditions Related to Genetic Changes

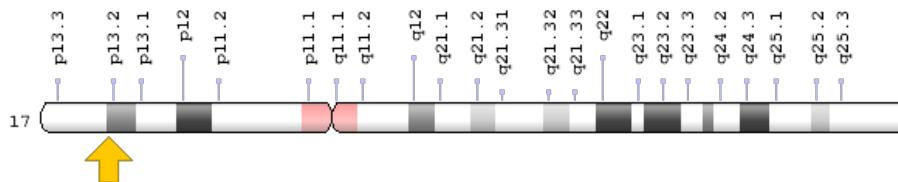
cystinosis

More than 80 different mutations that are responsible for causing cystinosis have been identified in the *CTNS* gene. The most common mutation is a deletion of a large part of the *CTNS* gene (sometimes referred to as the 57-kb deletion), resulting in the complete loss of cystinosin. This deletion is responsible for approximately 50 percent of cystinosis cases in people of European descent. Other mutations result in the production of an abnormally short protein that cannot carry out its normal transport function. Mutations that change very small regions of the *CTNS* gene may allow the transporter protein to retain some of its usual activity, resulting in a milder form of cystinosis.

Chromosomal Location

Cytogenetic Location: 17p13.2, which is the short (p) arm of chromosome 17 at position 13.2

Molecular Location: base pairs 3,636,391 to 3,663,103 on chromosome 17 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- CTNS-LSB
- CTNS_HUMAN
- Cystinosis
- PQLC4

Additional Information & Resources

GeneReviews

- Cystinosis
<https://www.ncbi.nlm.nih.gov/books/NBK1400>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28CTNS%5BTIAB%5D%29+OR+%28Cystinosis%5BTIAB%5D%29+AND+%28Genes%5BMH%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

OMIM

- CYSTINOSIN
<http://omim.org/entry/606272>

Research Resources

- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=CTNS%5Bgene%5D>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=2518
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/1497>
- UniProt
<http://www.uniprot.org/uniprot/O60931>

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Reprinted from Genetics Home Reference:

<https://ghr.nlm.nih.gov/gene/CTNS>

Reviewed: February 2008

Published: March 21, 2017

Lister Hill National Center for Biomedical Communications

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National Institutes of Health

Department of Health & Human Services